



## ADA2 gene

adenosine deaminase 2

### Normal Function

The *ADA2* gene provides instructions for making an enzyme called adenosine deaminase 2. This enzyme breaks down molecules called adenosine and 2'-deoxyadenosine. Because this enzyme functions in the spaces between cells, it is described as extracellular. Another form of adenosine deaminase, often called adenosine deaminase 1, breaks down the same molecules inside cells. This other version of the enzyme is produced from the *ADA* gene.

Researchers are still working to determine what roles the extracellular form of adenosine deaminase plays in the body. Studies suggest that it acts as a growth factor, which means that it stimulates cell growth and division. In particular, the enzyme appears to be involved in the growth and development of certain immune system cells, including macrophages, which are a type of white blood cell that plays a critical role in inflammation. Some macrophages are pro-inflammatory, meaning they promote inflammation, while others are anti-inflammatory, meaning they reduce inflammation. Adenosine deaminase 2 also seems to be essential for keeping the lining of blood vessel walls intact.

### Health Conditions Related to Genetic Changes

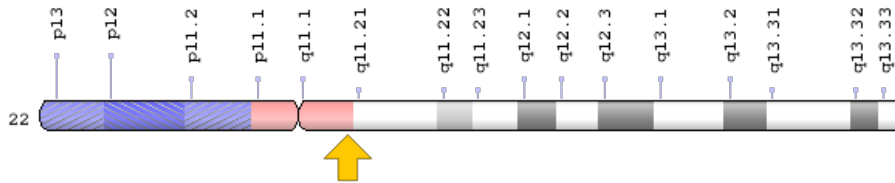
#### adenosine deaminase 2 deficiency

At least 15 mutations in the *ADA2* gene have been found to cause adenosine deaminase 2 deficiency, a disorder characterized by abnormal inflammation of various organs and tissues, particularly the blood vessels (vasculitis). These mutations severely reduce or eliminate the function of adenosine deaminase 2. Researchers do not fully understand how a loss of this enzyme's function leads to the features of ADA2 deficiency. They speculate that a lack of this enzyme may disrupt the balance between pro-inflammatory and anti-inflammatory macrophages in various tissues, leading to abnormal inflammation. The enzyme's role in maintaining the structural integrity of blood vessels could help explain why the blood vessels are most often affected by inflammation in this disorder.

## Chromosomal Location

Cytogenetic Location: 22q11.1, which is the long (q) arm of chromosome 22 at position 11.1

Molecular Location: base pairs 17,178,790 to 17,221,854 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- adenosine deaminase CECR1
- ADGF
- cat eye syndrome chromosome region, candidate 1
- cat eye syndrome critical region protein 1
- CECR1
- IDGFL
- SNEDS

## Additional Information & Resources

### Educational Resources

- Immunobiology (fifth edition, 2001): Neutrophils Make Up the First Wave of Cells that Cross the Blood Vessel Wall to Enter Inflammatory Sites  
<https://www.ncbi.nlm.nih.gov/books/NBK27122/#A213>
- Molecular Biology of the Cell (fourth edition, 2002): Blood Vessels and Endothelial Cells  
<https://www.ncbi.nlm.nih.gov/books/NBK26848/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CECR1%5BTIAB%5D%29+OR+%28adenosine+deaminase+2%5BTIAB%5D%29+OR+%28ADA2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- CAT EYE SYNDROME CHROMOSOME REGION, CANDIDATE 1  
<http://omim.org/entry/607575>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_CECR1.html](http://atlasgeneticsoncology.org/Genes/GC_CECR1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ADA2%5Bgene%5D>
- HGNC Gene Family: Adenosine deaminase family  
<http://www.genenames.org/cgi-bin/genefamilies/set/1302>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=1839](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1839)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/51816>
- UniProt  
<http://www.uniprot.org/uniprot/Q9NZK5>

### **Sources for This Summary**

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